### RARE DISEASES









# HISTORY doesn't have to REPEAT ITSELF GEN-XOME

generations GENETICS

### It's not just a diagnosis, it's hope!



A family history of a rare disease is **NOT** the kind of history that must repeat itself.

With more than **7,000** identified rare disease and approximately **80%** being linked to genetic causes, diagnosing rare diseases can be often take a long time – up to 7 years – with many financial & emotional burdens incurred. With **GEN**-XOME, this is not the case anymore!

#### **GEN-**XOME

#### An integrated service of caring

Powered by our extensive clinical experience, whole exome sequencing "GEN-XOME" unveils the root cause of rare diseases in a very quick and affordable way leading to better patient outcomes. Our team will be with you all the way from traveling back into your family history, up till opening new doors for almost certain diagnosis, treatment opportunities and even new healthy pregnancy plans.



### WORK WITH A TEAM **DEDICATED**



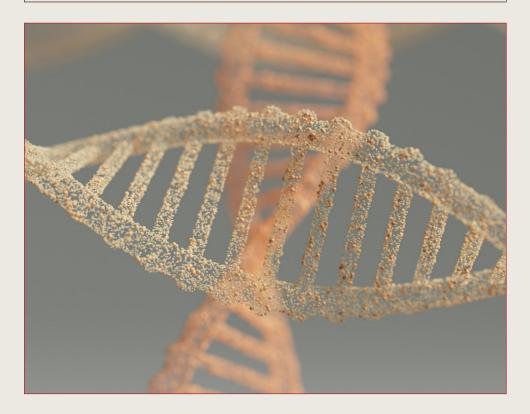
## TO IMPROVING PEOPLES LIVES

- **CLINICAL COUNSELING** WITH EGYPT'S ONLY SPECIALIZED GENETIC RARE DISEASES CLINICIANS
- 2 WORLD'S LATEST **SEQUENCING TECHNOLOGY** IN TESTING FOR
  RARE DISEASES
- 3 BIGGEST DATA-POOL OF THE EGYPTIAN POPULATION
- 4 RESULTS REVIEWED BY **AMERICAN BOARD CERTIFIED GENETICITS**
- 5 POST-TEST **FAMILY PLANNING**SESSION WITH VARIOUS TREATMENT
  & PREVENTION PLAN

# WHY GEN-XOME?

### The **SCIENCE** behind it

with new technologies in DNA sequencing it is now feasible to perform genome-wide sequencing on a routine basis.



In whole exome sequencing the **DNA** responsible for encoding proteins is sequenced. The exome, regions responsible for encoding proteins, consists of only **1-2% of a person's total genome** but harbors about **85%** of the causal variants identified in Mendelian disorders. This method allows for **detection of variants** in the coding regions of any gene and not just specific variants or specific genes.

### WHEN IS **WES**RECOMMENDED?



We recommend WES for complex & undiagnosed cases with suspicion of genetic causes.

WES is conventionally recommended when patients present complex, heterogeneous phenotypes that are suggestive of multiple conditions or are otherwise unclear or atypical. WES may also be recommended when a prior genetic test was unsuccessful. The latest clinical evidence also supports WES as a FIRST-LINE TEST when a patient's symptoms or family history suggests a genetic cause of the diseases.

This is especially true for **neurodevelopmental disorders**, including intellectual disability, global developmental delay, and autism spectrum disorder due to the high diagnostic yield. <sup>10, 13</sup> **The ACMG (American College of Medical Genetics and Genomics)** recommends the use of exome/genome sequencing as **FIRST-TIER TEST** for children with intellectual disability, developmental delay, or multiple congenital anomalies. <sup>14</sup>

The test results from **WES** may also lead to **more rapid** diagnoses, **improved** prevention of symptomatic illness, **more targeted** treatments or even end the need for some **costly or invasive** procedures.

#### generations genetics

A BETTER CHANCE AT LIFE